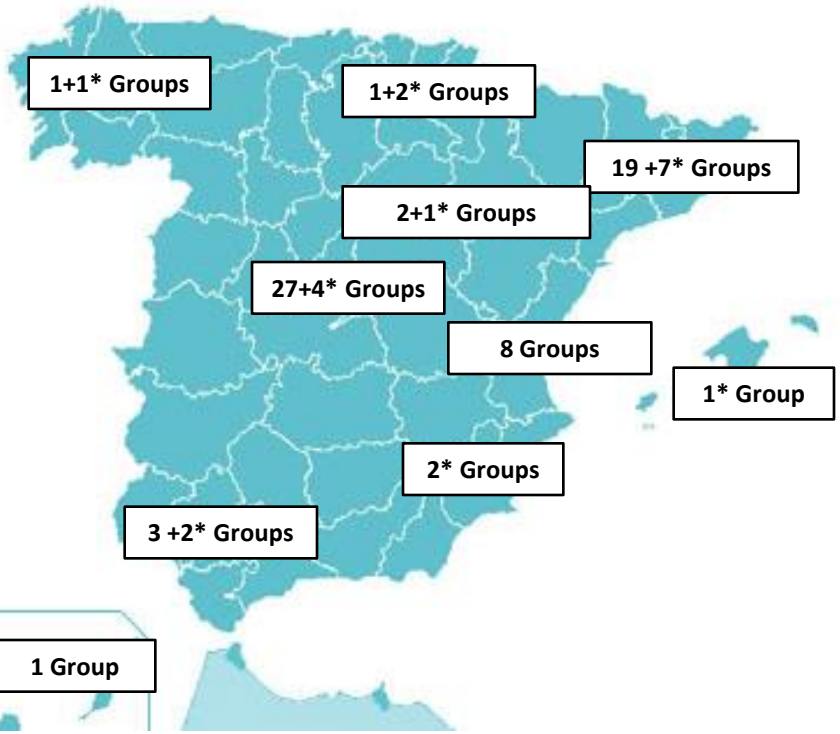
Centre for Biomedical Network Research on Rare Diseases

as a positive example of research initiative on RD in Spain

Dr. José M. Millán, PhD
Deputy Director of CIBERER
University Hospital La Fe, Valencia, Spain

What is CIBERER?

- **Spanish Consortium** creating and belonging to the Carlos III Institute of Health **(ISCIII)** - Ministry of the Economy and Competitiveness **(MINECO)** – since nov. 2006
- **CIBERER is a Innovative Network structure**
- **Is a reference centre in Spain for translational research into Rare Diseases.**
- Is a team of **over 700 basic biomedical scientists and clinical investigators** organized in:
  - **62 research groups**
  - **20 clinical associated groups***
  - **28 Spanish institutions as** Universities, Research Centres, hospitales, etc.



FEDER

EURORDIS
Rare Diseases Europe

Biotechnologies, Pharma, Hospitals, Universities, etc.

Patients, Associations, Society, Administrations

er

Cooperative Research

Network of excellence

Translational Research

From bench to bedside

62 research groups, multidisciplinary teams collaborating with hospitals, companies, patients, administrations at national & international level.

CIBERER´s groups are experts in

MNGIE, Retinosis Pigmentosa, Hirschsprung, Amaurosis congénita de Leber ,
Distrofia muscular de Duchenne y Becker, HHT, Allan-Herndon-Dudley,
cánceres hereditarios, síndrome de Temblor, Hipotiroidismo congénito, Defectos de diferenciación sexual,
Epidermolisis bullosa, Síndrome de Down, Distrofias de retina, Stargadt, Niemann-Pick C Sanfilippo, enfermedad de Danon,
Miopatias, Mc Ardle, defectos congénitos, Melanoma familiar, Retraso mental de origen genético FXTAS + X frágil,
Hipoacusias congénitas, Neurofibromatosis, Atrofia muscular espinal, Deficit de CoQ,
Enfermedades mitocondriales, Sindorme Wolfram, Cáncer de próstata familar, Cistinuria, Lisinuria,
Charcot-Marie-Tooth, Ataxias cerebelosas autosomica recesivas, Ataxia de Friedreich, Síndrome de Kindler,
síndrome de Werner, anemia de Fanconi, Tromboastenia de glanzmann, Síndrome de Bernard Soulier,
ELA, Leucodistrofias, Trastornos espectro autista, síndrome de Williams, Defectos congénitos,
Enfermedades lisosomales, MPS, Defectos de complemento, Enfermedades del ciclo de la urea,
Hiperoxalurias, Mohr-Tranebjaerg, Epilpsias monogenicas, Lafora, Enfermedades metabólicas hereditarias,
Acromegalia, Cushing, Ataxia de Friedreich, Enfermedades de Gaucher, hipercolesterolemia
familiar, Sindrome Adams-Oliver, Síndromes de sobrecrecimiento, Wilms ' tumors,
Síndrome Hemolítico-Urémico, Defectos de complemento, Angioedema hereditaria, Síndrome de Usher,
distrofias retinianas, Albinismo, Disqueratosis congénita, síndrome de Werner, Ataxias, Autismo, Adrenoleucodistrofia ligada
al X , síndrome de Ellis-Van Creveld, osteogénesis imperfecta,
Síndromes de mantenimiento del mtDNA, , **Progerias,** etc……

Internationalization Prog.

Human Research Progr

ER Training Progr.

Cooperative Research Projects Progr.

7 Scientific Programmes focus on groups of RD witch integrated the 82 research/clinical groups

Genetic Medicine	Inherited Metabolic Medicine	Mito. & Neuromuscular Medicine	Pediatric and Developmental Medicine	Neurosensorial Pathology	Endocrine Medicine	Hereditary Cancer and Related Syndromes

Working together on 4 thematic areas to foster translational research on RD

Genes, genomic medicine, bioinformatics and systems biology (includes research into clinical genetics)

Physiopathology of rare diseases

Clinical and epidemiological research into RD: towards Personalised Medicine

Innovation in Therapeutic research and advanced therapies in RD

& supported by several Transversal Programmes:

Support tools for scientific work

Platforms in RD

Communication Prog

Knowledge Transfer Prog.

How about CIBERER  ??

CIBERER receive funds:

- **ISCIII-MINECO (80 %)**
- through competitive projects at international /national levels (16%)
- private institutions/companies (4%)

CIBERER financed directly:

- **140 researchers** specialized on RD (PhD, Technicians, students)
- More than **100 Cooperative Project Research** on RD since 2006
- **Strategic Projects** (IRDiRC priorities)
  - **Non-diagnosed RD progr (Exomes NGS)**
  - **Spanish Exome Database - CIBERER Spanish Variant Server**
- **Scientifics Platforms** (Orphanet -Spain, Biobank, Bioinformatic platf., Service Phenotyping of Laboratory Animals on RD, etc)
- Workshops, Scientifics Meetings, Scientific & Social Newsletters
- Etc….

"walking together" Patients & Families + Researchers

- Celebrations of Annual **Rare Disease Day** *"Investigar es avanzar"* with patients associations

- **Collaborative Research Projects** with and thanks to patients associations: Lowe, DEBRA, OPITz, Sanfilippo, etc…

- Collaboration/supporting **Registries** : Wolfram, Fanconi Anemia, Neuromuscular, IIER national registry, etc.

- Collaboration and support of **joint patients-researchers events**: 22q11, HHT, Lowe, Albinism, Cerebrotendinous Xanthomatosis…

- **Institutional cooperation with FEDER**

International Collaborations

- **Orphanet- Spain:**

- **EUCERD JOINT ACTION:** Coordinator of WP about the Quality of Life and Expert Centres in close collaboration with the Spanish Ministry of Health, Social Services and Equality.

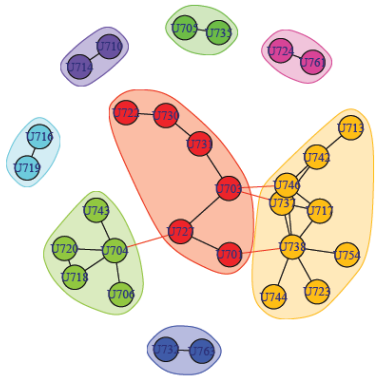
- **E-HOD - E-IMD:** European Network and Registry for Homocystinurias and Methylation Defects

- **RD CONNECT (associated to the project)** An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research

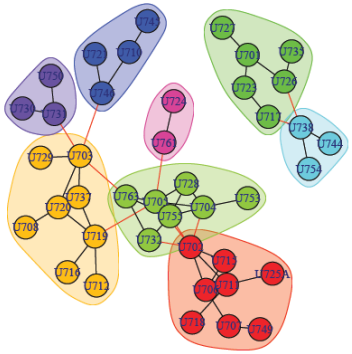
- Member of specifics committees/activities:
  - Fondation Maladies Rares (F. Palau)
  - IRDiRC (animal models, etc.)
  - EUROPLAN
  - Etc.

Results of CIBERER activities:
increase of the scientific collaboration and productivity by the CIBERER groups

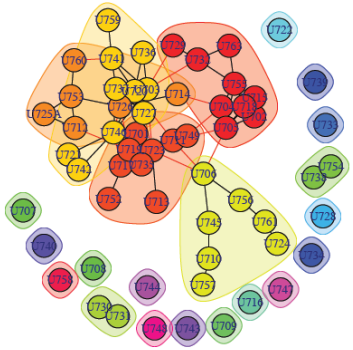
Evolution of intramural collaboration - CIBERER



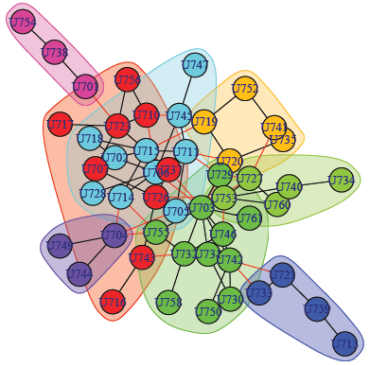
2000



2007

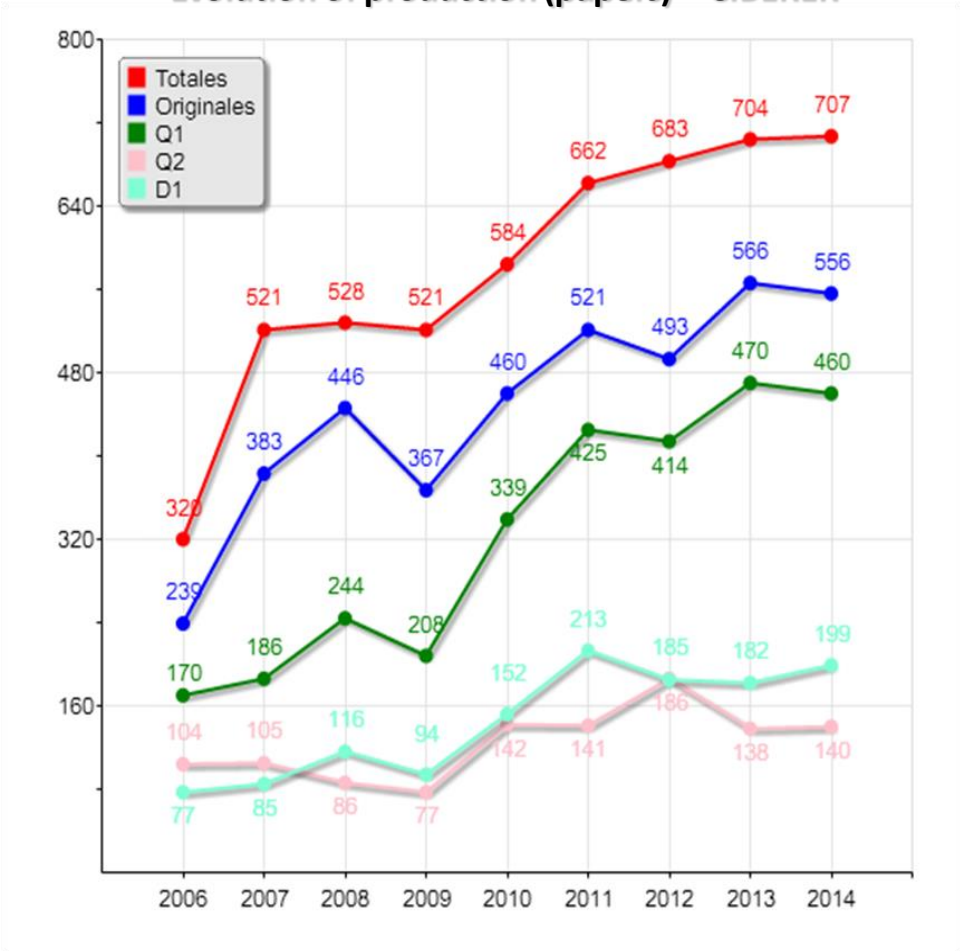


2010



2013

Evolution of production (papers) - CIBERER



CIBERER is contributing to the major IRDiRC overarching goals

Genic Therapy: Clincal trial on Fanconi Anemia…..

Designation of Orphan Drugs 4 (HHT, A. Fanconi, hiperoxaluria primaria, disqueratosis congénita, etc)

Creation y mantenance of registries on RD

Animal models for the study of RD: McArdle disease, X-linked adrenoleukodystrophy, CMT (Gdap1), Retinitis pigmentosa (Cerkl), Down syndrome (Dyrk1a, etc.), etc

Non-diagnosed RD progr => new genes (Gen NFU1: encephalopathy mitochondrial , Osteogenesis imperfecta, Fanconi Anemia, etc)

+ de 4.500 papers since 2.006, CIBERER is reference in the scientific production on RD at international & national level

ciberer

CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED
DE ENFERMEDADES RARAS

"A model of Cooperative Research on Rare Diseases"

More Information:

www.ciberer.es

info@ciberer.es

MINISTERIO
DE ECONOMÍA
Y COMPETITIVIDAD

Instituto
de Salud
Carlos III

Thank you very much for your attention!